Genetic Disease Program
Newborn Screening and Genetic Diseases Follow-up

The result of reducing staff or funding would impact the state's ability to test and follow-up on individuals with inheritable conditions. Louisiana Administrative Code 48: V.6303 mandates newborn screening and follow-up services for individuals with genetic conditions. Without the proper funding and staff, the debilitating effects of these conditions could not be prevented.

Objective

The objective of this activity is to ensure that all infants born in Louisiana receive a newborn screen and to eliminate or reduce mortality, morbidity and disabilities by early detection and treatment of the disorders included in the newborn screening panel.

Performance Indicators

1. Percent of infants born in Louisiana who received at least one newborn screen
2. To ascertain the number of infants with newborn screens
3. Percent of infants with a positive screen who received timely & appropriate treatment

Narrative

The Genetic Diseases Program, in collaboration with the State Public Health Laboratory, operates a statewide newborn heel stick screening and follow-up program in accordance with the pertaining legislation and rules: R.S. 40:1299.1.,2.,3.,4 and LAC 48: V. 6303. The newborn screening program started with screening for phenylketonuria (PKU) in 1964, and screening for 27 other diseases was added through the following years. The program's mission of early detection coupled with immediate medical management of an infant with one of these disorders prevents many, and in some disorders, all of the serious clinical sequelae.

<u>Newborn Heel Stick Screening and Follow-up</u>

The purpose of these services is to ensure the early detection and treatment of newborns with the diseases as listed on the newborn screening panel. In accordance with state law and rule (see state mandates above), all newborns in the state are screened before discharge from the hospital. Currently there are 28 diseases on the newborn screening panel which reflects the core panel recommended by the American College of Medical Genetics. The 10 most common

diseases listed on this panel are PKU, congenital hypothyroidism, biotinidase deficiency, galactosemia, sickle cell disease, argininosuccinic aciduria, citrullinemia, homcystinuria, maple syrup urine disease and medium chain acyl coA dehydrogenase deficiency. The symptoms associated with these diseases, when untreated, are mental retardation for PKU and congenital hypothyroidism, seizures and other neurological sequelae for biotinidase deficiency and galactosemia, and life-threatening infections in the first few years of life for sickle cell disease patients.

Clinical Genetics Services

The purpose of these services is to ensure that genetic evaluation and counseling is accessible to individuals in all regions of the state through the operation of genetics clinics at eight parish health unit sites, certain DHH hospitals, and the medical centers associated with the three medical schools. Providing an accurate diagnosis can be life saving for some patients as there may be treatment for the disorder.

Better Health

The program's mission of early detection coupled with immediate medical management of an infant with one of these disorders prevents many, and in some disorders, all of the serious clinical sequelae.

Newborn screening is an efficient and cost effective measure to ascertain any inborn errors of metabolism and other heritable conditions. The conditions on the newborn screening panel, if left untreated, can result in mental retardation, failure to thrive or even death.

As a preventive service newborn screening and genetics follow-up offers an opportunity for infants in this state to start life with the best health possible. It also addresses the issue of access to affordable and appropriate care. The 8 genetic clinics across the state allows for access to case management of the conditions on the screen and thereby reducing the need to access emergency rooms and providing access to patients in the more rural areas of the state.

- This activity is supported by the following state rules: RS 40:1299,l1,.2,.3; LAC 48: V.6303.8
- Supported by the United States Senate S.1858 – Newborn Screening Saves Lives Act of 2007
- For over 5 years, at least 95% of infants born in Lousiana have received at least one newborn screen. (MCH 2009 Title V Block Grant Data)
- For over 5 years, at least 95% of all infants with presumptive postivie results received timely and appropriate treatment (MCH 2009 Title V Block Grant Data)